

Hayes Genetic Test Experts can help you make defensible coverage determinations when members request specific genetic tests

Hayes

Transforming Healthcare with Evidence



Case Study 2

Coverage Policy Dilemma

The Hayes client received a request to perform a full analysis of the **4 genes** associated with Sanfilippo syndrome on all four members of a family who were at risk to be carriers of the defective gene. The price for each family member would be **more than \$6000**. The client needed expert advice to determine whether testing the entire family was the best approach.

Considerations

Within any given family, the genetic variant responsible for Sanfilippo syndrome is the same for all family members.

Hayes Solution

- The Hayes GTE Director recommended a **tiered approach to familial testing**, specifically that a comprehensive analysis should only be performed for one affected family member.
- If a variant is identified, the other family members can then be tested for that specific variant at a much **reduced cost**, usually **<\$500** per family member.
- If a variant is **NOT** found in the affected individual, then testing other family members would have **no benefit** given that the familial variant is unknown.
- Using this approach resulted in a **cost savings of at least \$5500** for each family member subsequently tested.

Disease Overview

Sanfilippo syndrome, also known as mucopolysaccharidosis type III, is an **inherited disease** of metabolism in which the body cannot break down long chains of sugar molecules.

Symptoms appear after the first year of life, and include gradually declining learning ability, below-average growth, and deteriorating mental status leading to several neurological impairment and retardation.

Sanfilippo syndrome is diagnosed in approximately **1 in 70,000** births. The syndrome is inherited as an autosomal recessive trait, meaning that both parents must pass on the defective gene.

Unbiased analyses of health technologies that support transparent, defensible coverage policies

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